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Abstract Topic:- Prenatal, perinatal and developmental genetics

Abstract Title:- Rapid Testing of G6PD Deficiency in Neonates for Clinical Settings

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Aims:-Birth defects constitute the 5th largest cause of death in newborns in India. Glucose-6-phosphate dehydrogenase deficiency (G-6-PDD) is a X-linked genetic disorder associated with increased risk of jaundice and kernicterus at birth. The present study aims to develop an indigenous rapid quantitative screening test for G6PDD

Methods:- G6PD detection test is based on fluorometric biochemical enzymatic determination of G6PD activity from specimens spotted on dried blood spots (DBS). The test was standardized using synthetic controls for formulating the reaction to obtain optimum results in short time and validated for analytical performance with LOD, linear range and precision. The clinical performance was compared with international platform of Labsystem Diagnostic.

Results:- The assay was developed for quantitation of G6PD within 30 minutes. The linear range covers concentrations from 0.2 U/gHb to 7.4 U/gHb. No cross reactivity was observed when tested with various hormones (galactose dehydrogenase, TSH, alkaline phosphatase) and metabolites (bilirubin, triglyceride, cholesterol). The precision was performed for repeatability and reproducibility with ≤20% coefficient of variation. The comparator clinical study showed 100 % concordance for deficient and sufficient DBS samples. The results were compared with commercially available "Varioskan Lux" microplate readers, and Mylab's Indigenous reader and the results were comparable.

Conclusions:- The study provides end to end integrated solution comprising test reagents and indigenous testing platform for G6PD screening. This test with its cost effective, fast, accessible and near POC utility will have a positive impact on mass screening of newborns in clinical settings and has been approved by CDSCO, India's regulatory body.

Keywords:- NBS, G6PD screening, DBS